Spontaneous Gestural Communication as a Predictor of Autism Spectrum Diagnosis in Children with Fragile X Syndrome.

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Abstract

This study aimed to determine if early spontaneous gestural communication is a predictor of later Autism Spectrum Disorder (ASD) diagnosis in children who have already been diagnosed with Fragile X Syndrome (FXS). The communication samples of 49 children were obtained across multiple contexts as part of a larger longitudinal study examining maternal responsivity. Videos were coded for communication form and function, and initiations of gestural communication were analyzed. There were significant differences between the two groups. The children in the FXS only group used distal points, proximal points, and representational gestures more often than children with FXS and ASD; however, children in the latter group were more likely to use give gestures. Overall, children with a single FXS diagnosis initiated more joint attention than the FXS + ASD group.

Dedication

This thesis is dedicated to my family and friends who have been so supportive of everything I do.

To Jeff and Lisa, my parents who always encouraged me to believe in myself and take every dream as far as it can go.

To my little brother, Daniel, the inspiration in everything I do.

And to my husband, Todd, thanks for always being patient, kind, and supportive on this (sometimes crazy) journey.

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Spontaneous Gestural Communication as a Predictor of Autism Spectrum Diagnosis in Children with Fragile X Syndrome.

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Introduction/Literature Review

Overview

This thesis focuses on reviewing potential early indicators of Autism Spectrum Disorder (ASD) and examining potential early indicators of ASD in children with Fragile X Syndrome (FXS). The first section of the literature review focuses on ASD and the current research on identifying early indicators of ASD in siblings of individuals with a diagnosis of ASD. The second section of the review discusses FXS and how individuals with FXS represent a high-risk population for ASD. Finally, the current study and research questions are introduced.

Autism Spectrum Disorder

Autism spectrum disorder (ASD) is a neurodevelopmental disorder that has the potential to impact many areas of both behavior and language. It is thought to be genetic in nature, but it is not yet known what causes ASD (Bailey, et al., 2004). Currently, the average age of diagnosis is around the age of three (Charman & Baird, 2002; Fombonne, 2005; Mandell, Novak, & Zubritsky, 2005); however, many parents report suspicions about atypical behavior before this time (Coonrod & Stone, 2004; Barbaro & Dissanayake, 2009). While the population of people with ASD is a largely heterogeneous group, the trademark symptoms are difficulties with social communication and restricted and repetitive behavior. Many other symptoms can be associated with ASD. These include intellectual disability, language delay, and other mental or behavioral

disorders such as anxiety and comorbid conditions such as Fragile X Syndrome (American Psychiatric Association, 2013).

An important topic in the ASD literature base is the discovery of traits and development of standardized measures that would allow for earlier identification of ASD. Identification at a younger age would allow for early intervention to take place which is ideal for this population. Harris and Handleman (2000) found that age and IQ at the time of intake into an intensive treatment program were predictive of later educational placement. Children who had a higher IQ and were a younger age at time of intake were the most likely to be in a general education classroom 4-6 years later while children with a lower IQ and older age at intake were more likely to be placed in a special education classroom. Landa and Kalb (2012) examined the long term effects of children with ASD who participated in a 6-month intensive intervention program at around 27 months. They found that gains in IQ and communication were present at 35, 41, and 72 month follow-ups. Research has shown that early intervention is correlated with school placement and developmental gains; however, it can sometimes be difficult to ascertain how large of a role language skills and/or IQ play when examining rate of skill acquisition (Dawson and Osterling, 1997). At this point, much of the research has been focused on finding key early indicators of ASD that can be utilized as part of an ASD assessment.

Sibling Studies and ASD

In general, research has shown that there are differences between children who are at a higher risk for ASD and children who are low risk for being diagnosed with ASD. Typically, children are categorized as "high-risk" if they have an older sibling that has been diagnosed with ASD. Some studies have examined siblings of children with ASD to look for early indicators of an ASD diagnosis. ASD occurs in siblings with an 18.7% occurrence rate suggesting a strong

genetic component (Ozonoff et al. 2011; Winder, Wozniak, Parladé, & Iverson, 2013). Twin studies have shown that even if a sibling does not have a diagnosis of ASD they are more likely to have language impairments and a broader autism phenotype (BAP) marked by difficulties with relationship building, impaired play and conversational skills, and mild stereotypy (Micali, Chakrabarti, & Fombonne, 2004; Le Couteur et al., 1996). By examining young children with a higher risk of being diagnosed with ASD, researchers are able to gain more information about the traits and early developmental patterns of individuals with ASD.

Ibañez, Grantz, & Messinger (2012) found that when they compared the developmental trajectory of referential communication in children with siblings diagnosed with ASD (high risk) and siblings with no family history of ASD (low risk) there were significant differences. Participants were examined across 8, 10, 12, 15, and 18 months of age. Nonverbal referential communication was defined in this study as the use of eye contact and gestures to communicate about objects with communication partners. The researchers looked at joint attention and behavior regulation. The child would get credit for initiations of joint attention (i.e., initiating the shared enjoyment or interest in an event or an object with the tester) and responding to joint attention (i.e., following a joint attention behavior of the tester, such as a point). Behavior regulation was also categorized as either initiating or responding. The high risk group showed less responsivity to bids for joint attention, and had a consistently lower level of joint attention initiation. However, the development of initiating joint attention followed a similar trajectory in both groups. That is, both groups had a period of growth followed by a brief period of decline before the trajectory increased again. While the groups appeared similar in initiation of behavioral regulation at the first time point the developmental trajectory of the group at higher risk for ASD showed slower growth than the low risk group's trajectory.

Clinicians are faced with the difficulty of wanting to provide early intervention to children with ASD, but there are not many standardized or gold-standard assessments that are available for diagnostic use with young children. Often even if an assessment states that it can be used for a range of ages including young children it may not be as valid when used with clients at the limits of the age range (Brian et al., 2008). Brian and colleagues (2008) followed a group of high-risk and low-risk infants to find out if current gold-standard ASD assessments and an assessment they created could differentiate between children with ASD and those without at 18 months. The groups were assessed at 18 months and then again by an examiner blind to the previous results at 36 months. After the 36 month assessment, the groups were divided into 3 groups: high-risk with ASD, high-risk without ASD, and low-risk. They found that the Autism Diagnostic Observation Schedule (ADOS) module 1 and Autism Observation Scale for Infants (AOSI) given at 18-months both had many individual questions/items that differentiated between the ASD and non-ASD groups. The areas from the ADOS included items from the social domain, communication domain, and behavioral domain. The AOSI showed similar differences in the groups as well as had unique domains that differentiated the ASD and non-ASD groups. These were transitions, motor control, and reactivity. One limitation of this study is that the results come from analysis of the group as a whole rather than at the individual level which is more representative of how a child would be diagnosed. Furthermore, the researchers were looking to see if the results of the 18-month assessment predicted the diagnosis at 36-months. The diagnosis was not given at 18-months.

There are a few screening tools for ASD that have been studied for use in young children. Rowberry et al. (2014) examined the First Year Inventory (Baranek et al. 2003), a parent report screener, with high-risk and low risk groups. The screener consists of 61 questions involving

questions on a 1-4 Likert scale and multiple choice questions that attempt to assess the areas of social communication and sensory regulation. The examiners found that the children who were in the high-risk group and received a diagnosis of ASD were rated differently from their other high-risk peers by both clinicians and parents. The researchers suggested that by using a combined score of the social communication and sensory regulation sections the assessment should be able to differentiate between high-risk children without ASD and high-risk children with ASD. However, there were some limitations to this study. The researchers suggested that the parents of the high-risk group (who already had one child diagnosed with ASD) may rate their children differently than parents without children with ASD due to their knowledge and experience. Additionally, the purpose of a screener is to identify those who need additional testing, and there is a limited amount of additional testing that can be done at that age.

The Modified Checklist for Autism in Toddlers (MCHAT; Robins, Fein, Barton, & Green, 2001) and Social Communication Questionnaire (SCQ; Rutter, Bailey, & Lord 2003) are commonly used to screen young children who show signs of ASD. Robins, Fein, Barton & Green (2001) found that the MCHAT was able to accurately classify 33 of 38 children between 18-30 months with ASD into the correct category. Some studies have found that the SCQ has low sensitivity when using the recommended cut off score of 15 with young children under 44 months (Wiggins, Bakeman, Adamson, & Robins, 2007).

Gesture Differences

Children with ASD often show delays across various language areas. Mitchell et al. (2006) hypothesized that language and communication skill delays could be predictive of later ASD diagnosis in children with an older sibling previously diagnosed with ASD. In Mitchell et al. (2006), the children within the high risk group who ultimately received a diagnosis of ASD at

24 months understood fewer phrases and produced fewer gestures at 12 months as measured by the MacArthur Communicative Development Inventory (Fenson, et al., 1993) than the low risk group and the children in the high risk group who did not receive a diagnosis of ASD. The MacArthur however does not provide a thorough assessment of gesture use and therefore does not serve as a reliable measure of gestures (only 12 gestures are included and can only be rated as 'not yet,' 'sometimes' or 'never), and as a parent report measure is subject to potential bias. At 12 months the children in the high risk group without ASD did not differ significantly from the controls on the MacArthur; however, at 18 months these children were producing fewer gestures and producing and understanding fewer words than the low risk group. This suggests that receptive language and gestures might provide a reliable means of differentiating between not only children who are high risk from those who are low risk for ASD by around 18 months, but also can differentiate between high risk children without ASD and high risk children with ASD by one year of age.

Winder, Wozniak, Parladé, & Iverson (2013) also conducted a study involving young children with older siblings diagnosed with ASD (high risk group) and children with no family history of ASD (low risk group). These participants were videotaped and the videos were coded for spontaneous communication attempts and mode of communication (vocalizations, gestures, etc.). This methodology provides a more valid means of comparing groups than standardized parent measures such as the MacArthur as it removes the factor of potential parental bias (Rowberry et al., 2014). This study found that high risk infants had in general less spontaneous communication, produced less communicative vocalizations and words, and produced less gestures. Of the 15 high risk children, 3 ultimately received a diagnosis of ASD. These 3 children stood out in the high risk group in that they produced no words, fewer communicative

vocalizations, and limited gestures when compared to the rest of the high risk group. This study, like Mitchell et al. (2004), suggests that communicative gestures and vocalizations could be an early indicator able to differentiate between high-risk children who are later diagnosed with ASD and high-risk children who do not receive a diagnosis.

Communicative Function

It has been fairly well documented in the literature that children with ASD are likely to exhibit deficits in joint attention skills (Maljaars, Noens, Jansen, Scholte, & Berckelaer-Onnes, 2001; Chiang, Soong, Lin, & Rogers, 2008). Chiang, Soong, Lin, and Rogers (2008) examined the communication of 28 children with ASD (23-40 months of age), 24 with developmental disabilities (18-43 months of age), and 27 younger typically developing children (13-15 months of age) and 25 older children (18-20 months of age). The researchers found that the children in the ASD group had less nonverbal communication, significantly fewer proximal points, and less initiation of and response to joint attention. The group of children with ASD also made fewer requests. Maljaars and colleagues (2011) found similar results when they examined the communication of 26 3-11 year old children with ASD and 26 children who were typically developing and matched by mental age to the participants in the ASD group. They found that the children in the ASD group communicated less frequently than their mental age matched typically developing peers and tended to use primarily behavior regulating communication functions rather than social interaction or joint attention.

Fragile X Syndrome

Fragile X Syndrome (FXS) is also considered a condition that is at high risk for co-morbid autism (Budimirovic & Kaufmann, 2011). FXS is caused by a mutation of the FMR1

gene that results in an expansion of a CGG trinucleotide sequence on the X chromosome (Verkerk et al. 1991; Hagerman, 2008). Due to the mutation's location on the X chromosome, males are typically affected more frequently and more severely than females (Abbeduto, Brady, & Kover, 2007; Hagerman, 2008). As many as 30% of individuals with Fragile X Syndrome (FXS) have a co-morbid diagnosis of autism spectrum disorder (ASD) (Denmark, Feldman, & Holden, 2003; Kaufmann et al., 2004; Philofsky, Hepburn, Hayes, Hagerman, & Rogers, 2004), and FXS makes up 4-5% of ASD cases (Fisch, 1992). FXS is the most common inherited cause of developmental disability (Crawford, Acuna, & Sherman, 2001; Sherman, Morton, Jacobs, & Turner, 1984; Turner, Webb, Wake, & Robinson, 1996), and FXS occurs in approximately 1 in 4,000 males and 1 in 6,000 females (Centers for Disease Control and Prevention, 2011). The average age of diagnosis of Fragile X is around 35-36 months for boys and around 41 months for girls (Bailey, Raspa, Bishop, and Holiday, 2009).

In terms of the behavioral phenotype of FXS, there is a wide variety of strengths and weaknesses, and FXS can manifest itself in a range of disabilities from learning disabilities to severe intellectual disabilities (Loesch, Huggins, & Hagerman, 2004). Boys with FXS generally have intellectual disability falling within the moderate to severe range (Hagerman, 2002; Hooper, Hatton, Baranek, Roberts, & Bailey, 2000). Girls with FXS generally have abilities ranging from mild intellectual disability to normal intellectual functioning (Cronister, Hagerman, Wittenberger, & Amiri, 1991). Areas of relative strength for individuals with FXS include receptive and expressive vocabulary (Abbeduto et al., 2003). Areas of weakness include social communication (Murphy & Abbeduto, 2003), morphosyntax (Abbeduto & Hagerman, 1997), and adaptive behavior (Fisch, Simensen, & Schroer, 2002). Additionally, boys with FXS tend to be less intelligible than their peers and have delayed speech sound acquisition (Barnes et al.,

2009; Roberts et al., 2005). Autism spectrum disorders (ASD) can be a comorbid diagnosis with FXS, and this will be discussed in more detail below. However, even without a diagnosis of autism, individuals with FXS have symptoms similar to those found in autism spectrum disorders (Bailey et al., 2004; Hall, Lightbody, Hirt, Rezvani, & Reiss, 2010). These can include impairments in language, difficulties with social interactions, stereotyped behaviors and restricted interests (Feinstein & Reiss, 1998; Bailey et al. 2000; Abbeduto, Brady, & Kover, 2007). This overlap in symptomology can make it difficult to differentiate between having one, the other, or both. In some clinics, as soon as a child is diagnosed with ASD the diagnosing team will suggest testing for FXS.

The emergence of expressive language is often delayed in individuals with FXS, and while initially the parents usually notice delays in both receptive and expressive communication, by about 30 months most parents believe that expressive language is more of a concern than receptive language (Roberts, Hatton, & Bailey, 2001). Receptive language is a relative strength for children with FXS, and studies have shown that their receptive vocabulary doesn't differ significantly from children of a similar mental age (Roberts et al., 2007).

FXS and ASD

Since children with FXS represent a population at elevated risk for ASD (Denmark, Feldman, & Holden, 2003; Kaufmann et al., 2004; Philofsky, Hepburn, Hayes, Hagerman, & Rogers, 2004) a similar approach to the sibling studies mentioned above could be utilized to find differences between children with FXS alone and children with FXS and a co-morbid diagnosis of ASD. Many of the sibling studies discussed in this paper found differences in high risk siblings with and without ASD diagnoses by examining their communication development and use. It makes sense that communication differences could also differentiate between children

with FXS alone and FXS with ASD. Also, while FXS and ASD can be co-morbid diagnoses, there are not many evidence-based ways to differentiate between those who have FXS alone and those who have FXS and ASD diagnoses (Bailey et al., 2004).

Current Study

The goal of this study is twofold. The first is to investigate further the idea that the frequency and type of early spontaneous communication can differentiate high risk children who will receive a diagnosis of ASD from high risk children who don't as seen in Winder, Wozniak, Parladé, & Iverson (2013). This paper will primarily focus on the early use of spontaneous gestures. The second goal is to add to the growing number of studies that have been able to document the communication differences between children who only have FXS and children who have FXS and ASD. The current study will examine the number and type of spontaneous gestures of children with FXS and determine whether this differentiates children with FXS who end up having a co-morbid diagnosis of ASD from the children who do not end up with a diagnosis of ASD.

Research Questions

1. Are there significant differences in the types of early spontaneous gestures observed in children with FXS who are later diagnosed with autism, compared to children with FXS who do not receive this diagnosis?
    a. Hypothesis: Children in the FXS only group will use a wider variety of gestural communication than children in the FXS and ASD group.
2. Are there significant differences between the aforementioned groups in the function of these early spontaneous gestures?

a. Hypothesis: Children with FXS only will be more likely to use both joint attention and behavior regulation while children with FXS and ASD will be more likely to use primarily behavior regulation.

Methodology

Participants

Participants for this study were 49 children with full mutation FXS. All of the participants were part of a longitudinal study looking at family adaption to FXS and the effects of maternal responsivity on development (For more information see Brady et al., 2014; Warren et al., 2010). Families were recruited from a variety of races, family backgrounds and socioeconomic statuses. The participants in this study were between the ages of 15-36 months of age at the time of the gestural assessment. The group consists of 38 boys and 11 girls. 15 of the participants met the study criteria for having a dual diagnosis of ASD. 12 of the participants had scores on the CARS that were within the autism range at the age of 9 years 6 months and also had scores above the cut-off for at least 2 other observations (hence they were consistently above cut-off for ASD according to the CARS.) 2 had scores at least 4 points above the cut-off at the time of the recording (between 15-36 months of age), but did not have data for the later time points. 1 participant did not have a score above 30 at the age of 9 years 6 months, but was very close to the cut off and had scored above the cutoff at previous time points.

Procedures

The participants received home visits from graduate research assistants. During these home visits the child was videotaped during different contexts around the home (see Table 1). During the book context, the mother and child were provided with books by the examiners to

read or they could use their own books. They were videotaped for 5 minutes while they read or talked about the book. The free play context consisted of the mother playing with her child for 5 minutes. The naturalistic context is taken from a 30 minute video of the mother and child performing typical daily tasks ranging from household chores to playing. Two 5 minute clips were selected from this video to make the naturalistic 1 and naturalistic 2 contexts. Finally, the snack context consisted of the mother and child making a snack together. Snack items were provided by the examiners or the participants could use their own materials. The dyad would be filmed for 5 minutes during this final activity.

Because of the range of ages in the participant group, the videos used were either taken from the first time point of the longitudinal study or the second time point to include all participants when they were at least 15 months of age (mean CA=29.1months, range= 15 months – 36 months).

Coding

All of the videotaped contexts were coded previously for communicative form and function for both the mother and child. This included whether the child initiated a communicative attempt, and the form and function of the communication attempt.

Communication forms and functions. Forms of communication included words, vocalizations, gestures, pointing, and signs, as well as combinations of these forms.

Functions included joint attention, behavior regulation, and behavior compliance. The mother's communication was coded as well. The videos were coded using Noldus' The Observer 10 software. Multiple graduate research assistants were trained in the coding system and

reliability was calculated for 31% of the videos. For more information about how these were coded see Brady et al. (2014) and Warren et al. (2010).

New Coding for the Current Study. For the purpose of this study, codes pertaining to gesture type were added to the existing coding scheme. Gestures were divided into two categories: deictic gestures and representational gestures. Deictic gestures are gestures that are used to draw attention to or request an object, person, place, or event (e.g., pointing, showing, giving, etc.) Representational gestures are gestures that demonstrate a particular concept such as shrugging, shushing, and waving (Winder, Wozniak, Parladé, & Iverson, 2013). These two categories were further divided into individual gesture codes, and the exact gesture type was coded. Deictic gestures included reaches to request, gives, show gestures, proximal points, and distal points. Representational gestures included waves, nods, shrugs, shushes, and high fives. (For exact definitions of the gestures see Table 2.) (Hahn, Zimmer, Brady, Swinburne Romine, & Fleming, 2014; Winder, Wozniak, Parladé, & Iverson, 2013; Brady, Fleming, Thiemann-Bourque, Olswang, Dowden, & Saunders, 2012).

The gestures were coded by a graduate student after training by the first author. Reliability coding was completed by another graduate student trained for the current study, and who demonstrated adequate reliability with the primary coder. The training of the reliability coder consisted of reading a coding manual created for this study and meeting with the primary coder to discuss definitions. During the meetings the primary coder also showed examples of the various kinds of gestures being coded. The criteria for reliability was completing the videos of a participant not included in the study across all the same contexts with an average of 80% reliability or above for both gesture type and gesture function.

After the reliability coder met training criterion, 14/53 files were independently coded by both the primary coder and the reliability coder. The overall mean percent agreement was 87.2%. Percent agreement was calculated by comparing each instance of a gesture code, totaling the number of matching codes, and dividing by the total number of gesture codes. In some files there were only one or two gestures which made it difficult to achieve a reliable percentage agreement if the coders disagreed on a gesture. The coders would review items that were coded differently, but did not conduct consensus scoring. See Table 3 for percent agreement for each individual category.

Childhood Autism Rating Scale (CARS). The CARS (Schopler, Reicher & Renner, 1988) was used to measure the frequency and severity of autistic behaviors. The CARS has 15 items and the total score has a range of 15 to 60. Scores at or above 30 indicate possible autism and scores above 36 indicate possible severe autism. The CARS has established criterion-related validity (r = .84). According to the test manual (Schopler, Reicher & Renner, 1988), the CARS was found to be valid across alternate conditions and with a professionals from varied disciplines. The CARS has strong internal consistency reliability (.94), test-retest reliability (.88), and good interrater reliability (.71).

To obtain CARS scores, two examiners who had experience working with children including those with developmental disabilities were trained to administer the CARS with 80% agreement. Both examiners would independently complete the CARS during observation of the participant, and would compare scores after the observation had taken place. If any discrepancies arose, they would discuss them and arrive at a consensus for the score.

The participants of the study were determined to have ASD or not based upon their CARS scores, and the stability of the CARS score over time (how consistently the score was

above or below the score of 30). If a participant consistently had a CARS score above 30 they would be considered to have ASD and if they consistently had a score below 30 they did not. This variable was used to group the participants into the FXS only group or the FXS + ASD group.

Analysis. The data were obtained from the previously described gesture coding system and included the number of each gesture type used to initiate communication and the function of those gestures. The early initiation gestures and function data were examined to determine if the amount of initiation at an early age predicts later diagnosis of ASD. The CARS scores were used to separate the participants into ASD and non-ASD groups. ANOVAs were run with follow-up comparisons between the two groups to find significant group effects if they existed.

Results

Data reduction. The analyses focused on gestures with at least 20 occurrences, and high reliability (at least 80% agreement). The gestures that met this criteria were distal points, proximal points, gives, and representational gestures.

The representational gesture category was reduced as follows. Waves, nods, shrugs, and shushes were combined into one category because each of these gestures occurred infrequently. Claps were removed from the representational gesture data because it was difficult to determine if claps were representational gestures (representing excitement/celebration), or if they were part of a social game or stereotypical behaviors. High fives never occurred.

Additionally, we did not analyze results for reaches, push away gestures, and shows because it was often difficult to determine if these gestures were intentionally communicative. For example, children often reached to an object in an effort to directly obtain the object. These

reaches would not be considered as intentionally communicative unless the child also looked up toward the examiner.

Differences in Gesture types. First, we compared differences in gesture use across children later diagnosed with ASD to those who did not meet criteria for ASD using a one-way ANOVA. There was a significant interaction between autism status and different gesture types at the p<.05 level ($F\ (3,141) = 3.28$, $p = 0.047$, *partial* $\eta^2 = 0.07$). This interaction indicated that gesture frequencies differed significantly across the two groups. Based on this result, we conducted additional follow-up comparisons to examine how each gesture type differed across groups. Significance was measured at the $p < .05$ level. There was no significant difference between the groups in terms of gives; however, individuals in the ASD group did produce more give ($M = 2.20$, $SD = 3.30$) gestures than individuals with FXS only ($M = 1.59$, $SD = 1.73$), $t = -0.68$, $p = 0.51$, $d = 0.24$. Participants in the non-ASD group produced more distal points ($M = 1.15$, $SD = 1.71$) than participants in the ASD group ($M = 0.04$, $SD = 1.05$), $t\ (41.52) = 1.87$, $p = 0.07$, $d = 0.80$. The difference in distal points was not statistically significant but the p-value of 0.07 and Cohen's *d* equaling 0.80 suggests that the difference is large and that the difference may be significant with a larger sample.

Participants in the non-ASD group produced significantly more proximal points ($M = 2.88$, $SD = 5.03$) than participants in the non-ASD group ($M = 0.27$, $SD = 1.03$), $t\ (38.76) = 2.89$, $p = 0.006$, $d = 0.86$. Representational gestures were also used significantly more frequently by participants in the non-ASD group ($M = 0.76$, $SD = 1.35$) than those in the FXS + ASD group ($M = 0.07$, $SD = 0.26$), $t\ (38.10) = 2.90$, $p = 0.006$, $d = 0.86$. Figure 1 shows the mean rates of different gesture types for the two groups.

Differences in gesture functions. Following this analysis, functions of each gesture type were analyzed. Table 4 shows the percentage of each function by each gesture type across both groups of participants. To determine if there were significant differences between the groups with regards to their use of the function types, t-tests were used to analyze differences in the percentages of each function type used in each group. The results demonstrated that there was a significant difference between the two groups. Participants in the FXS only group used more joint attention ($M = 0.63$, $SD = 0.34$) than the FXS + ASD group ($M = 0.25$, $SD = 0.36$), $t\ (14.96) = 3.07$, $p = 0.004$, $d = 1.11$.

Discussion

The purpose of this study was to examine the differences in early spontaneous gestural communication in children with FXS to determine if early gestural use is a predictor of later co-morbid diagnosis of ASD. Results indicated that there are significant differences between the two groups both in terms of form and function of gestural communication. Children who belonged to the FXS only group used significantly more proximal points, and representational gestures, and more distal points. Children in the FXS group used gestures primarily for joint attention, with the exception of gives. Gives were used to communicate behavior regulation and joint attention approximately equally. Children in the FXS + ASD group used distal points for both joint attention and behavior regulation and gives primarily for behavior regulation. This supports research findings that individuals with ASD are less likely to produce joint attention than both typically developing peers and peers with other developmental disabilities (Chaing, Soong, Lin, & Rogers, 2008). It is difficult to make a determination about the trends regarding function of representation gestures and distal points for with the ASD + FXS

group as each of these gestures only occurred a few times in the data each by only one participant. In general, these results support the current research that is available. Studies like the ones mentioned above in the introduction have shown that children with ASD initiate communication less, primarily use behavior regulation, and have a smaller communication repertoire than their typically developing peers and peers with other developmental disabilities (Chaing, Soong, Lin, & Rogers, 2008; Ibañez, Grantz, & Messinger, 2012; Winder, Wozniak, Parladé, & Iverson, 2013).

Implications of this study. One implication of this study is that the frequency and form of early gestural communication could be predictive of ASD in children with FXS and possibly for other children as well. This supports findings that early gestural communication can differentiate between individuals with ASD and individuals who are considered high-risk but ultimately do not receive an ASD diagnosis (e.g., Winder, Wozniak, Parladé, & Iverson's (2013) comparison of high-risk infants and low-risk infants). This information could inform future research on assessment measures that could be used with younger children to diagnose ASD earlier than the current average age of 3. While newer gold-standard ASD measures might be a ways off, this information in the meantime can inform clinicians to early red flags that a child might need to be tested for ASD. They could find out through information gathered during other assessments such as the ADOS (Lord, Rutter, DiLavore, & Risi, 1999) and Communication Complexity Scale (Brady et al., 2012), observation, or parent report whether the child is using a variety of gestural forms and functions in addition to other assessment materials to determine if further testing for ASD is needed.

Another implication of this study is that it demonstrates a behavioral difference between children with FXS and children with FXS and ASD. Many of the features of FXS overlap with the symptomology of ASD and it can be difficult to separate the two diagnostically. The results of this study add to the literature base about diagnostic differences between FXS and ASD. Even though FXS and ASD have similar symptoms, they differ in expected ways. For example, Chaing, Soong, Lin & Rogers (2008) found that individuals with ASD produce less joint attention than individuals with other developmental diagnoses. The participants with ASD in this study produced less joint attention than the participants with FXS only. This information can be used to create more sensitive measures that effectively parse out individuals with FXS from those with FXS + ASD so that appropriate intervention and care can be provided.

Finally, this information can help to guide clinicians in their intervention with individuals with FXS and individuals with FXS + ASD. An appropriate goal for an individual with FXS + ASD might be to increase gesture use or target joint attention skills while these goals may be less appropriate for individuals with FXS alone.

Limitations of the present study. One limitation of this study is the number of participants that fell into the FXS + ASD category. Only 15 of the 49 participants matched the study criteria for a dual diagnosis. By having so few participants fall into the FXS + ASD category, the results have limited generalizability to the entire population of individuals with ASD and FXS. Additionally, it means that the confidence interval for the measures of the FXS + ASD group is larger and therefore we cannot be as confident about the measures obtained. Finally, with a larger sample size variables like frequency of distal points may be statistically significant.

The contexts used for the study might have had some effect on the kinds of communication encouraged or discouraged. For example, the book context might have encouraged more responses than initiations or the snack context might encourage more behavior regulation than joint attention. This could mean that the data represents a more restricted view of how the child would actually communicate on a day to day basis in normal everyday situations.

Future Directions. Future directions from this study are to examine spontaneous communication more fully in terms of vocalizations, verbalizations, and combinations of communication forms to determine if they have similar differences to what was found in this study. This would be useful for identifying a more complete profile of communicative differences associated with later diagnosis of FXS or FXS + ASD. Information from such studies could be used to further delineate the diagnostic differences between ASD and FXS as well as inform clinicians about possible targets for intervention when working with individuals with these diagnoses. Additionally, examining these differences longitudinally would help to determine the developmental trajectory of gesture use, and whether or not the differences change over time.

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Appendix

Table 1.

Contexts of videos.

Context	Description	Length of time
Book	Mother and child read a book	5 minutes
Free Play	Mother and child played together	5 minutes
Naturalistic	Mother and child performed daily tasks and routines.	10 minutes
Snack	Mother and child made and ate a snack.	5 minutes

Table 2.

Definitions for gesture coding

Category	Gesture	Definition
Deictic Gestures	Reach	Child reaches one or both hands forward to request an action or object.
	Give	Child gives the parent an object either for the purpose of drawing attention to the object or requesting an action.
	Show	Child shows the parent an object by holding it up towards the parent.
	Proximal Point	Child's index finger is extended towards a referent with all other fingers at least slightly curled. The index finger should be closer than 6 inches to the referent.
	Distal Point	Child's index finger is extended towards a referent will all other fingers at least slightly curled. The index finger should be at least 6 inches away from the referent.
	Push Away	Child pushes away person or object to indicate rejection or refusal.
Representational Gestures	Wave	Child either waves 'hello' or 'goodbye'.
	Nod/Shake	Child nods or shakes head to indicate 'yes' or 'no'.
	Shrug	Child shrugs shoulders by moving shoulders towards ears indicating uncertainty.
	Shush	Child raises fingers to lips indicating a request for quiet.
	High Five	Child raises hand to request a high five.

Table 3.

Percent Agreement for Individual Codes

Code	Percent Agreement
Reach	84% (54/64)
Give	86% (36/42)
Show	61% (22/36)
Proximal Point	93% (62/67)
Distal Point	100% (23/23)
Push Away	74% (43/58)
Representational Gestures	82% (18/22)

Table 4.

Functions of Gesture Types.

Group		Non-ASD			ASD	
Gesture Type	Total #	% BR	%JA	Total #	%BR	%JA
Distal point	54	37	63	6	50	50
Proximal point	98	6	94	4	0	100
Gives	54	55	45	33	82	15
Representational	26	38	63	1	100	0
Marginal Means			63			25

Figure 1. Means of Gestures across Groups.